



# *X-Plain*™

## Amniocentesis

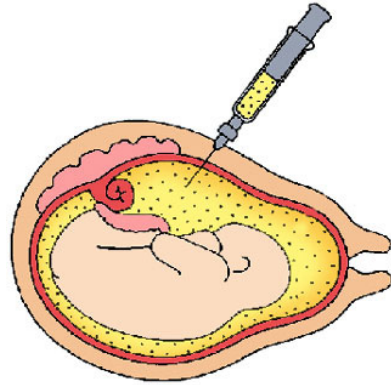
### Reference Summary

#### Introduction

Amniocentesis is a medical test to check the liquid that surrounds the fetus. It helps the doctor make sure the fetus is healthy.

For amniocentesis, the doctor inserts a needle into the uterus through the abdomen. It is a safe procedure. However, there are some risks.

If you are thinking about having an amniocentesis, this reference summary will help you decide based on the benefits and risks of the procedure. You will learn about when and why the doctor may recommend an amniocentesis. This reference summary also reviews alternative tests, how the procedure is performed, the risks involved and what to expect afterward.



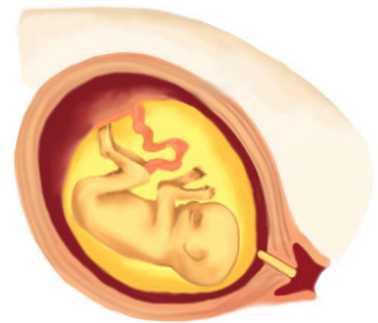
#### Pregnancy

In order to understand amniocentesis, it is important to know about certain parts of the body and how they work. This section talks about the amniotic sac, genetic diseases and pregnancy.

A fetus gets nutrition through the placenta. The placenta is a special organ attached to the wall of the uterus during pregnancy. The fetus' blood gets oxygen and nutrients from its mother's blood through the placenta.

As well as delivering oxygen and nutrients, the mother's blood picks up waste from the fetus' blood as it moves through the placenta. The fetus' blood travels to the placenta through the umbilical cord.

The fetus floats in a liquid called amniotic fluid. The fetus and amniotic fluid are in the amniotic sac. Waste products and cells that the fetus sheds are in the amniotic fluid.



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Like all body cells, amniotic fluid cells each have a nucleus. The fetus' chromosomes are in the nuclei of amniotic cells. Chromosomes are like the "instruction manual" for building the body. They control the way the body develops, including inherited traits.

There are 23 pairs of chromosomes. There are thousands of genes on each chromosome. Genes define the looks and some of the health conditions of a person. This is called the fetus' genetic material. All cells of a person have the same genetic material. Therefore, checking amniotic fluid cells is just like checking the fetus' cells.

Chromosomes can be studied to find out if the fetus has a genetic disease. A genetic disease occurs if a chromosome is abnormal. Sometimes a single abnormal gene on a chromosome causes a disease.

An example of a genetic disease due to an abnormal chromosome is Down Syndrome. Most people have two #21 chromosomes but people with Down syndrome have three #21's. People with Down Syndrome have mental and physical retardation.

Duchenne muscular dystrophy, on the other hand, is caused by an abnormality in one gene on the 23rd chromosome. It causes severe muscular weakness that leads to death by age 20.

## **Indications**

Amniocentesis is mainly done to check for genetic defects in a baby.

Amniocentesis can also be used late in pregnancy to find out whether the baby's lungs are ready for life outside the uterus. This is usually done if the baby needs to be delivered before term.

Another reason for amniocentesis is to check for infections in the uterus and amniotic fluid. Sometimes the doctor is concerned about these types of infections in pregnant women.

Amniocentesis can be used to check for differences between the mother's blood and the baby's blood. This is called Rh sensitization. The Rh, or Rhesus, antigen is a chemical found on the surface of red blood cells in some people.

Amniocentesis is safe. There is a 0.5% risk, or 1 in 200, of having a miscarriage. For pregnant women who are younger than 35 and healthy, the risk of having a baby with a genetic disease is so small that amniocentesis is usually not recommended.

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Doctors recommend amniocentesis for pregnant woman 35 and older because at that age, the risk of the baby having genetic defects is higher than the risk of amniocentesis. Doctors also recommend amniocentesis for women younger than 35 who are carriers of a genetic disease or have a family history of genetic disease.

If there is only a very small amount of amniotic fluid or if the placenta is at the front of the uterus, amniocentesis cannot be done. An ultrasound before amniocentesis helps check for these situations.

Amniocentesis is usually not recommended before the 14th week of pregnancy. Before the 14th week, the risks are higher and the results are not as accurate.



### **Alternatives**

Pregnant women older than 35 may have concerns about fetal abnormalities. There are a couple other test options besides amniocentesis. One is called the Quadmarker and another is called the CVS test.

The Quadmarker is a blood test that measures 4 substances in the blood. This test does not have risks. However, it does not identify birth defects as accurately as amniocentesis.

Quadmarker is good at checking for trisomy 21 and trisomy 18. Babies with either of these genetic diseases have 3 rather than 2 #21 or #18 chromosomes, respectively. Other genetic abnormalities are not detected with a Quadmarker. The doctor usually recommends amniocentesis if a Quadmarker test comes back positive.

CVS stands for chorionic villus sampling. It is not available in many places. A sample of placenta tissue is taken through the vagina or the abdomen. The test can be done 6 weeks earlier than amniocentesis, around 10 to 12 weeks into the pregnancy. However, it has twice the risk of miscarriage and does not identify as many diseases as amniocentesis.

A new blood test and ultrasound is now available to check for Down Syndrome, some other genetic diseases and some congenital heart defects. It can be done during the first trimester, between 11 and 14 weeks into a pregnancy.

Some parents choose the option of not testing for fetus abnormalities at all.

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Parents have different reasons for wanting to know about their child's health. Some plan to terminate the pregnancy if the fetus is seriously unhealthy. Others want to prepare for the challenge. For certain medical conditions, the fetus can be treated during pregnancy.

## Procedure

An obstetrician performs the amniocentesis in a doctor's office or at a hospital. It is an outpatient procedure, which means you can go home after the test. It takes about 15 to 20 minutes. There are no special requirements before the test.

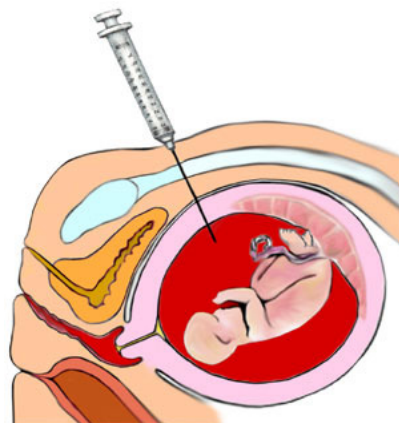
You will be asked to urinate before the test. Next, you will need to take off your clothes below the waist. There will be a sheet for you to wrap around your waist. You will lie on your back with your upper body slightly raised.

First, the doctor uses an ultrasound to find out where the fetus, placenta and amniotic fluid are. An ultrasound helps the doctor to know the safest place to insert the needle.

The obstetrician cleans your lower abdomen with antiseptic and you will get a shot of medication to numb the skin where the test needle will be inserted. You will feel a sharp, stinging sensation for a few seconds.

The needle is inserted through the abdomen and into the uterus, without hurting the fetus or the placenta. It feels similar to when you have blood drawn.

About 2 tablespoons of amniotic fluid are withdrawn into the syringe attached to the needle. To stay comfortable during this process, breathe slowly and relax the muscles of your abdomen.



After the fluid is withdrawn, the needle is removed and the puncture site is covered with a bandage.

After an amniocentesis, you may feel a little weak or nauseated. You may also have mild abdominal cramps.

You should not do anything strenuous for an hour after an amniocentesis. After about an hour, you can go on with normal activities unless your doctor tells you otherwise.

Notify your doctor immediately if you develop any of the following symptoms.

- Moderate to severe abdominal pain or cramping

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- Chills or a fever higher than 100 °F or 38 °C
- Dizziness
- Bleeding or fluid leakage from your vagina or from where the needle was inserted
- Redness or swelling where the needle was inserted
- Call your doctor if you notice any change in the fetus' activity.

## Risks

Amniocentesis is usually very safe. There is a 0.5% chance, or about 1 in 200, that the procedure may cause a miscarriage.

There is a risk of bleeding or fluid leakage from the vagina. These problems only occur in about 1% of women who have amniocentesis. Doctors monitor these complications, and if they do not resolve on their own, they can be treated.

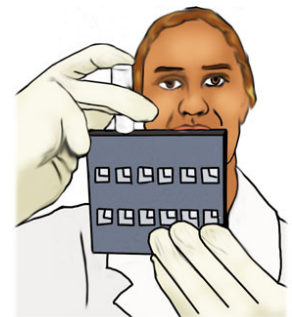
Rarely, the fetus is poked with the needle during an amniocentesis. This is usually harmless. The fetus floats away from the needle when it gets close to it.

Amniocentesis creates a very small risk of bleeding that could lead to the mother's blood mixing with the fetus' blood. Therefore, a pregnant woman with Rh– blood is given a vaccine to prevent Rh sensitization.

Results from amniocentesis that come back “normal” do not guarantee that the baby is healthy. Some defects, such as cleft palate, do not show up with amniocentesis.

In some cases, amniocentesis is recommended before the 14th week of pregnancy. In such cases, the risk of miscarriage is higher and the genetic results are not as accurate. There is also a risk of limb deformity.

After amniocentesis, the sample of amniotic fluid is sent to a lab for analysis. Results usually take 10 days to 3 weeks, depending on the lab. It takes that long to get results because the cells have to grow in the lab in order to analyze them.



In the lab, genetic and chemical tests are done. For genetic tests, certain chromosomes and genes are analyzed. For chemical tests, proteins, minerals and other compounds in the amniotic fluid are analyzed.

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If the results of an amniocentesis are “abnormal” a genetic counselor can discuss your choices with you.

## **Conclusion**

Amniocentesis is a safe procedure that can provide helpful information about the health of a fetus. It may be offered to women who are at a higher risk of having a baby with a genetic disease.

The risk of miscarriage for amniocentesis is about 0.5%, or 1 in 200. It is your decision whether the benefits of the procedure outweigh the risks.

This health education program gave you basic information to help you make an informed decision. Ask your doctor any questions you may have about amniocentesis.

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